RADY 416 CASE PRESENTATION

Amisha

INTERNATIONAL VISITING MEDICAL STUDENT MAY 2018

Edited by John Lilly, MD

History and Examination

- A 34 year-old male presented with complaints of cough and green sputum since the last 7 days. He also complains of dyspnea and fatigue
- He has a past history of recurrent respiratory infections, sinusitis and ear infections
- No history of smoking, fever, hemoptysis, chest pain, no family history
- On examination, bilateral basal wheezing was observed.

What is the next step in management?

ACR Appropriateness Criteria[5]

Clinical Condition: Acute Respiratory Illness in Immunocompetent Patients

Variant 4: Younger than age 40 and positive physical examination or other risk factors.

Radiologic Procedure	Rating	Comments	RRL*
X-ray chest	9		☢
CT chest without IV contrast	4		☢ ☢ ☢
CT chest with IV contrast	3		☢ ☢ ☢
CT chest without and with IV contrast	1		☢ ☢ ☢

Rating Scale: 1,2,3 Usually not appropriate; 4,5,6 May be appropriate; 7,8,9 Usually appropriate

*Relative Radiation Level

What is the next step in management?

Plain radiograph of the chest:- POSTERO-ANTERIOR (PA) and LATERAL views

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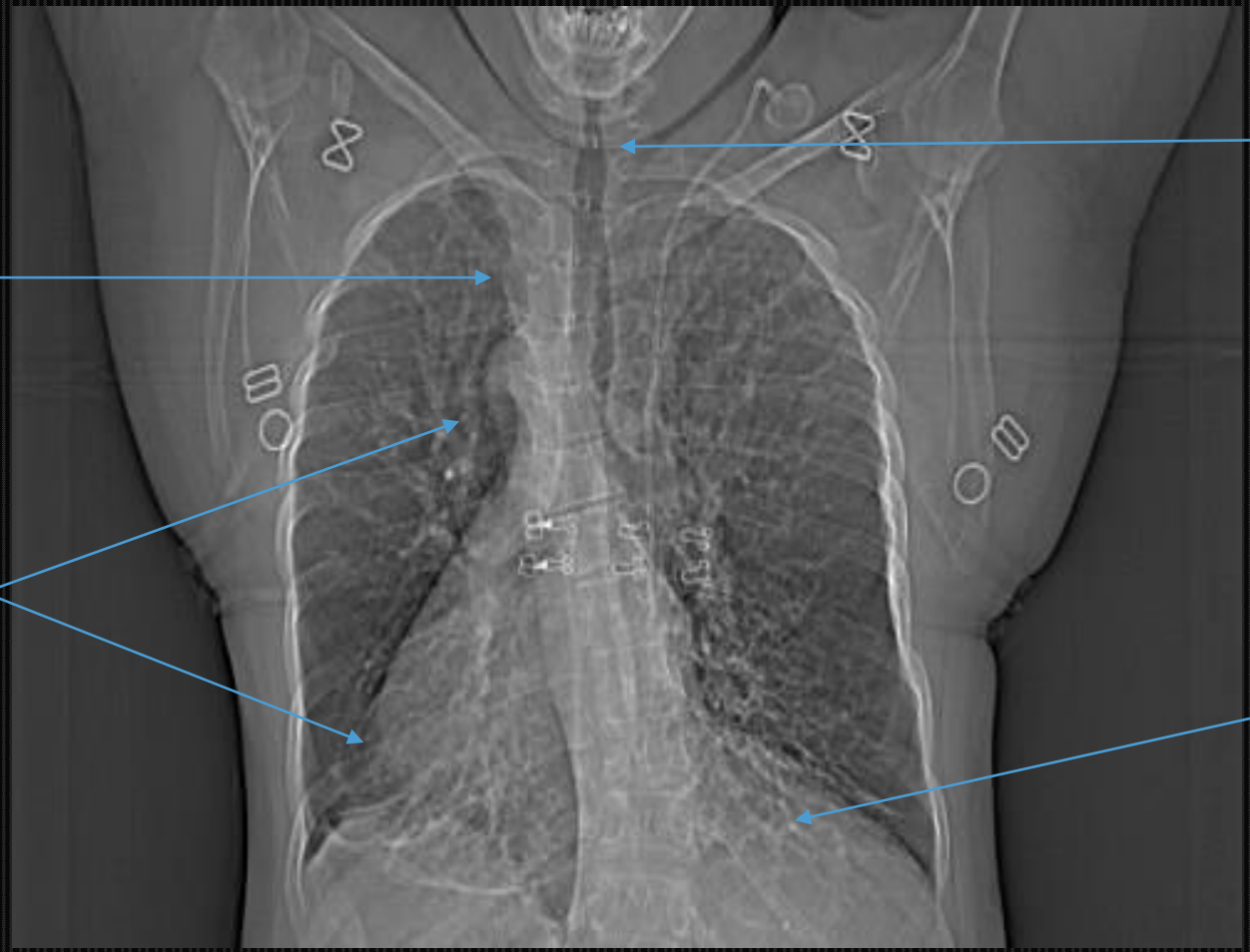
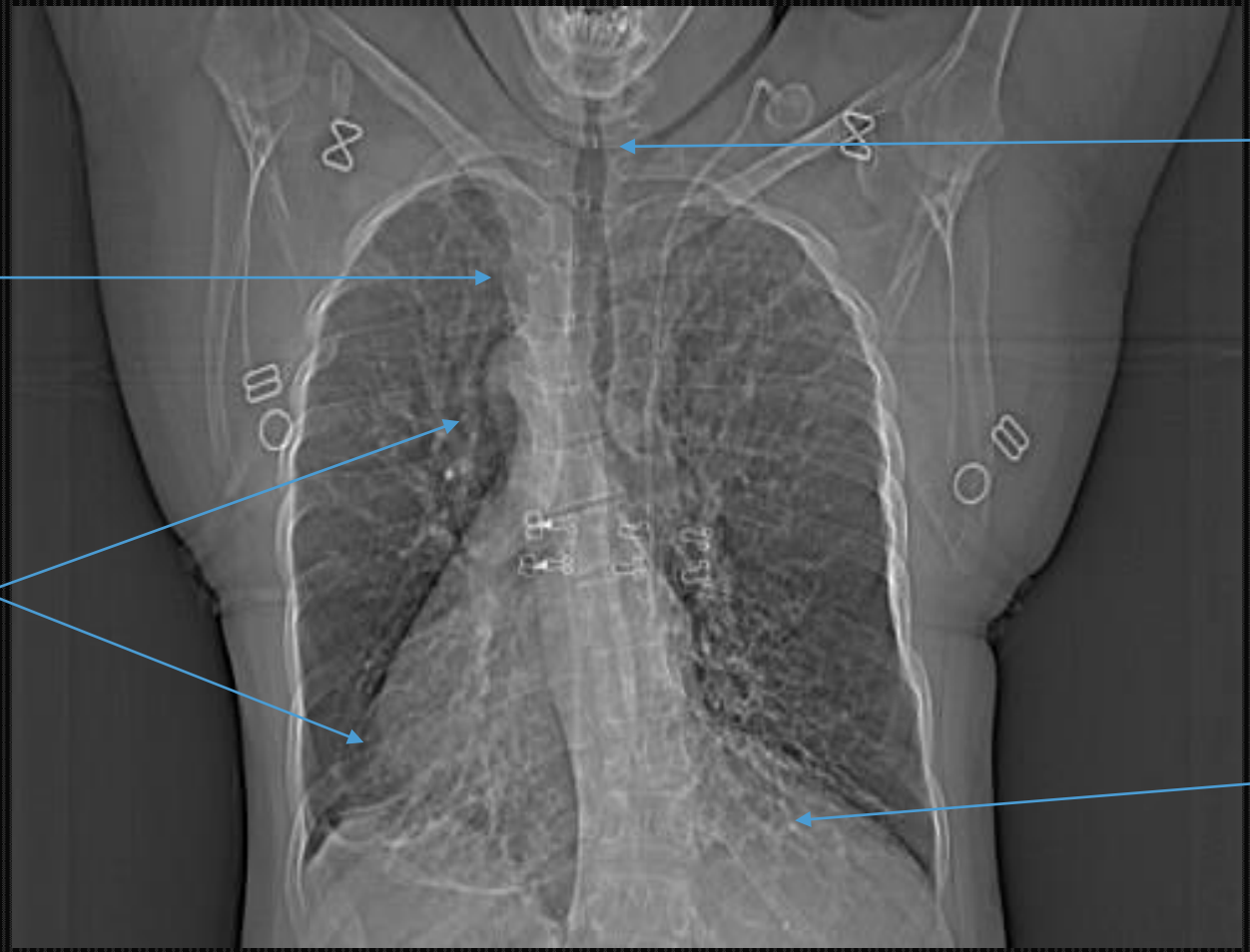
Imaging: PA CXR . . . Findings?



Imaging: PA CXR . . . Findings!



Midline Trachea

Mild Scoliosis

Aortic knob and Cardiac apex on the right (Dextrocardia)

Dilated bronchi and bronchial wall thickenings in bilateral basal lungs

What was the next step in management?

Complicated pneumonia prompted CT scan as next test (ours was ordered with IV contrast)

ACR Appropriateness Criteria[5]

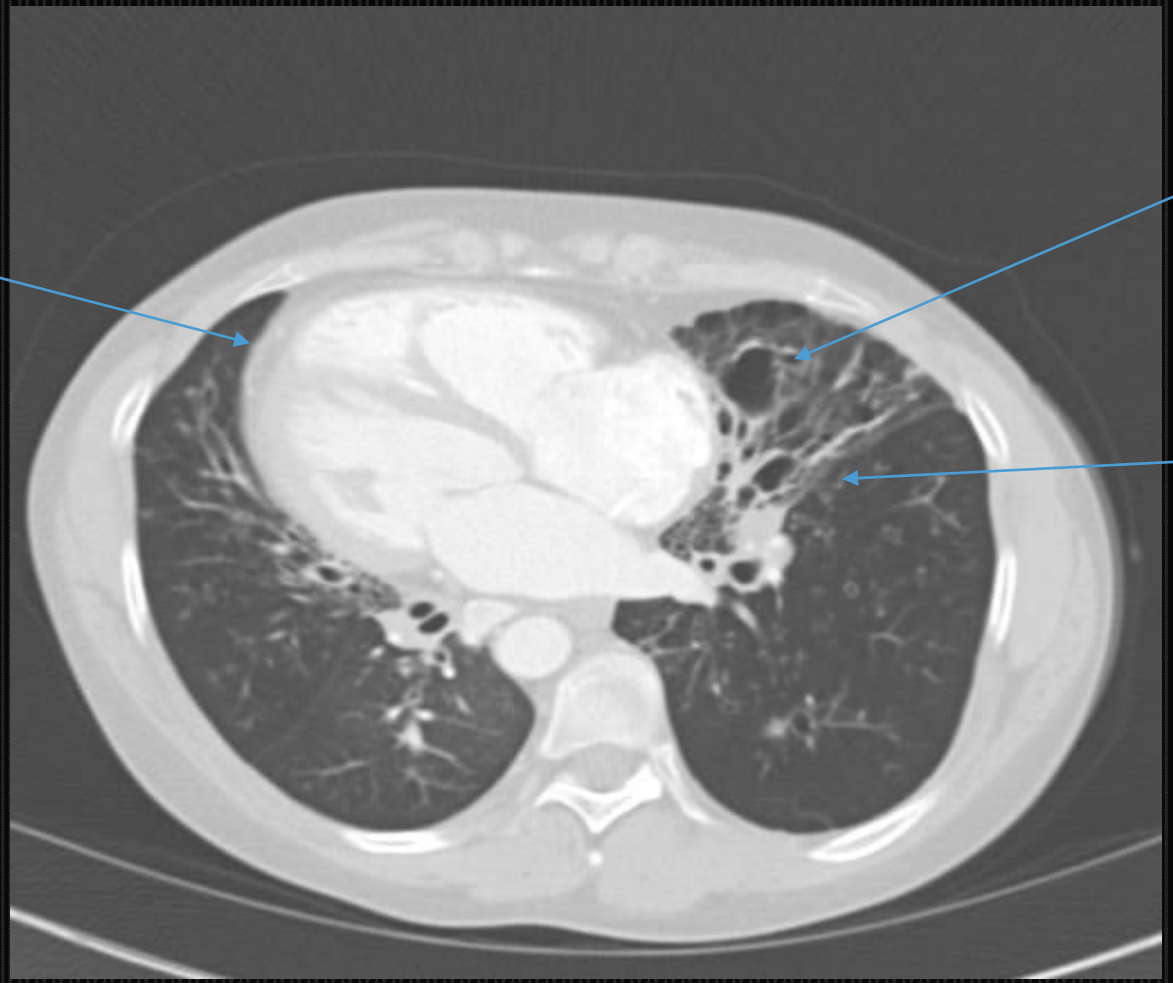
Variant 5: → **Complicated pneumonia.**

Radiologic Procedure	Rating	Comments	RRL*
X-ray chest	9		☢
CT chest without IV contrast →	8	If pneumonia is not resolving or intervention is contemplated. →	☢ ☢ ☢
CT chest with IV contrast	5		☢ ☢ ☢
CT chest without and with IV contrast	2		☢ ☢ ☢

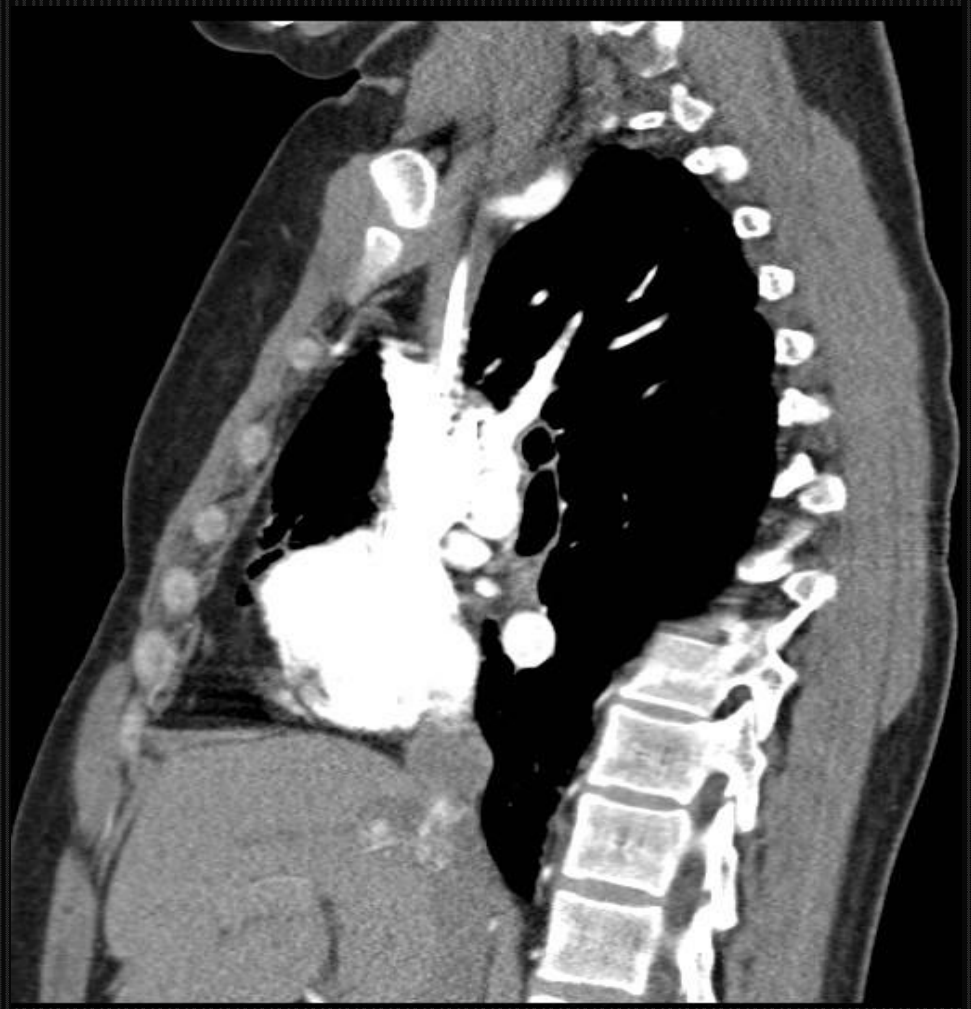
Rating Scale: 1,2,3 Usually not appropriate; 4,5,6 May be appropriate; 7,8,9 Usually appropriate

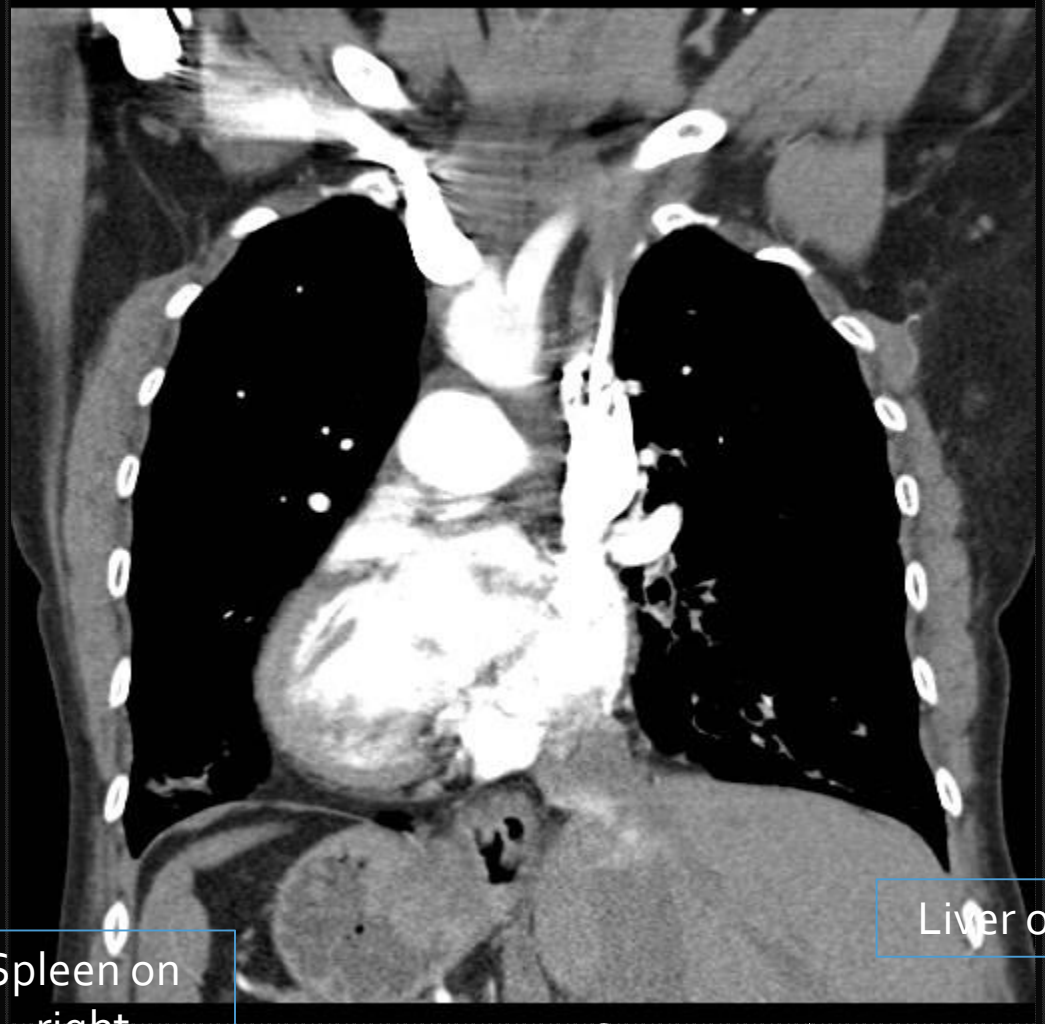
*Relative Radiation Level

Imaging: CT Chest transverse view



Imaging: CT Chest sagittal and coronal views



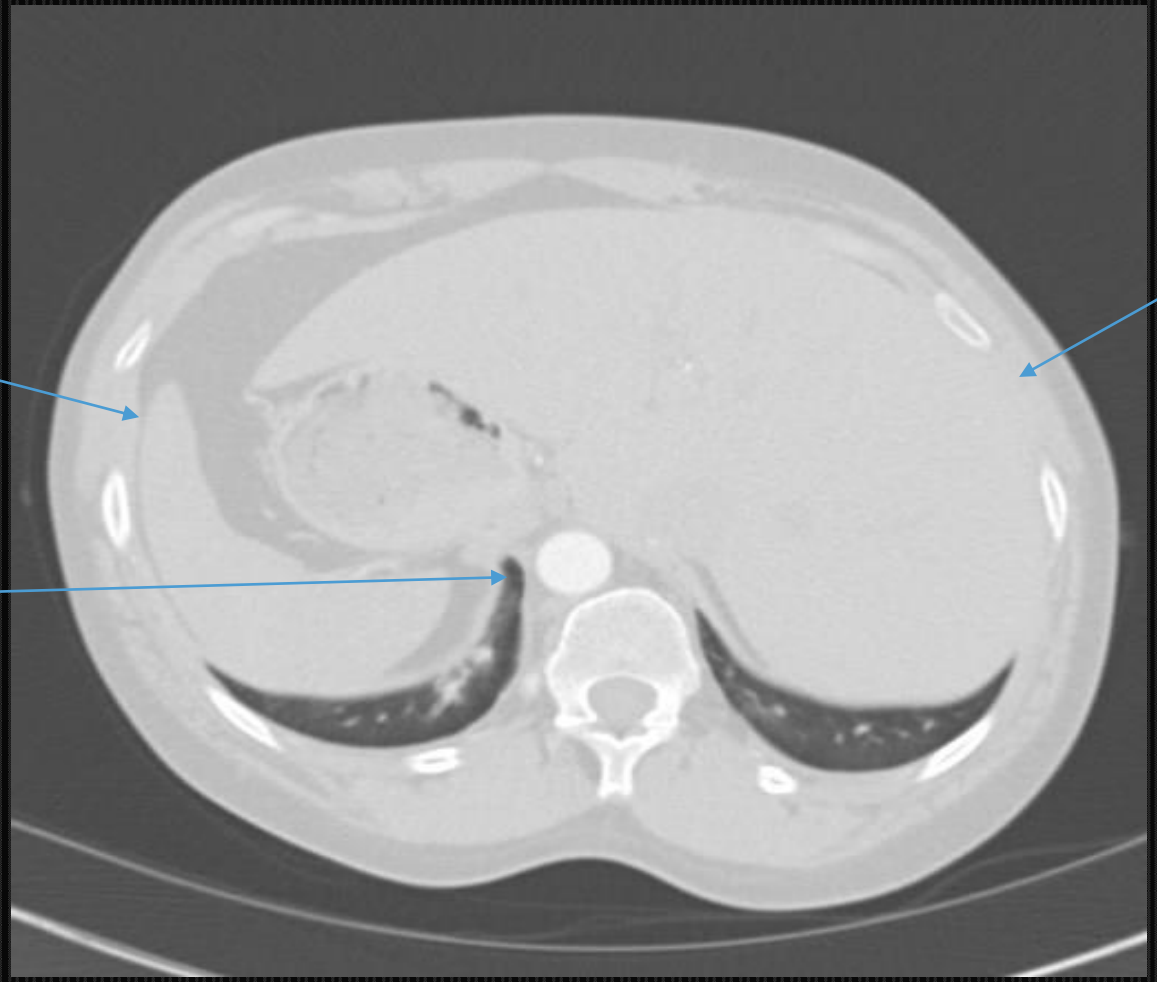


Spleen on right

Liver on left

Imaging: CT Chest transverse view

SITUS INVERSUS TOTALIS



Diagnosis

- Based on imaging modalities, a preliminary diagnosis of Kartagener Syndrome was made (Bronchiectasis, Sinusitis, situs inversus totalis)[1,5]
- Diagnosis should be confirmed by confirmatory tests: Transmission electron microscopy (TEM) analysis in nasal/bronchial brush biopsy or ciliary beat pattern (CBP)/ciliary beat frequency (CBF) analysis[1].

Diagnosis: Primary Ciliary Dyskinesia (PCD)

Autosomal recessive inherited disorder that includes a heterogeneous group of ultrastructural defects involving the cilia, resulting in their reduced or disorganized beating

The term 'immotile cilia syndrome' may be a misnomer as some movement may be present, although it lacks coordination and strength

Pathology[1]

- Movement of cilia is also important in organ placement in the developing embryo.
- Approximately 50% of individuals with PCD have Kartagener syndrome in which the internal organs including the heart, liver, spleen and intestine are on the opposite side of the body (situs inversus totalis).
- Some individuals with PCD have a condition called Heterotaxy (situs ambiguus) in which internal organs are abnormally positioned and have abnormal structure[4].

Clinical Features[1,4]

- Early-onset refractory or recurrent infections of the upper and lower respiratory tracts; which may include episodes of otitis media, mastoiditis, sinusitis, bronchitis and pneumonia
- Bronchiectasis is a common sequela of PCD, typically involving the dependent zones, including the lower lobes, right middle lobe, or the lingular segments of the left upper lobe
- Male infertility and ectopic pregnancy in females

Associations[4]

Syndromic

- Kartagener Syndrome (Sinusitis, Bronchiectasis, Situs inversus totalis
- Young Syndrome (Rhinosinusitis, Infertility, Bronchiectasis)

Non-syndromic

- Pectus Excavatum
- Hydrocephalus: thought to be due to abnormal ependymal cilia
- Congenital heart disease
- Biliary Atresia

Diagnosis[1]

CONFIRMATORY TESTS

- Ciliary beat frequency (CBF)/ciliary beat pattern (CBP): abnormal, reduced, or absent ciliary motion
- Transmission electron microscopy (TEM): Ultrastructural analysis of respiratory cilia demonstrating absence of, or abnormal dynein arms

SCREENING TESTS

- Saccharin test: time taken for a sweet taste to be appreciated in the mouth, when a saccharin pellet is placed on the inferior turbinate, normal = less than 30 minutes
- Nasal nitric oxide measurement: Typically very low (<250 ppb) in patients with PCD, CF, and severe rhinosinusitis

Imaging PCD Across Modalities

CXR:

Bronchial wall thickening

Bronchiectasis

Hyperinflation and
in some instances, cystic bronchiectasis with air-fluid levels may be visible

HIGH RESOLUTION CHEST CT (HRCT):

Most striking pulmonary abnormality is bronchial wall thickening and bronchiectasis, present in most patients

The distribution is either central or diffuse and has a predilection for the lower and middle lobes

PARANASAL SINUS CT:

Evidence of chronic sinusitis

May occur as early as 6 mos

Middle ears are opacified

20% have sinonasal polyps

Patient Treatment

- The primary goal of therapy is the prevention of bronchiectasis
- Chest physiotherapy assists in the clearance of mucus
- Antibiotics should be prescribed in case of evidence of infection
- Bronchodilators can be used if symptomatic wheezing is present, or for documentation of reversible airway obstruction
- Prevention of lung infection by measles, pertussis, influenza, and pneumococcal vaccines is highly recommended
- Additional preventive measures include avoidance of cigarette smoke and other airway irritants.

Differential Diagnosis[4]

- Immunoglobulin deficiencies (which also predispose to recurrent infection)
- Cystic fibrosis : Upper lobe predominance, and even when the lower lobes are involved, the disease is more severe in the upper lobes
- Chronic indolent infection such as Mycobacterium Avium Complex: middle lobe and lingula of middle-aged females
- Chronic recurrent aspiration
- Allergic bronchopulmonary aspergillosis : Central mid-lung zonal distribution of bronchiectasis

PCD vs CF: Distinguishing the 2

SIMILARITIES

- Both are hereditary with autosomal recessive inheritance
- Both have sinus and lung disease
- Both have infertility in males

- Primary ciliary dyskinesia
  - Basically normal mucus, cilia cannot move normally
  - Normal ductus deferens, sperm cannot swim normally
  - Radiologic findings are much milder

- Cystic Fibrosis
  - Abnormal mucus, cilia cannot clear it
  - Normal sperm, ductus deferens is obliterated
  - Radiologic findings are more severe

References

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